HIRSUTISM IN CHILDREN: PITFALLS AND DIAGNOSTIC CHALLENGES

Teofana Otilia Bizerea-Moga [1,2], Raluca Corina Tămășanu [1,2], Giorgiana Brad [1,2], Alexandra Velcelean [2] , Otilia Mărginean [1,2]

[1] "Victor Babeș" University of Medicine and Pharmacy, Timișoara, România
[2] "Louis Țurcanu" Children's Clinical and Emergency Hospital, Timișoara, România

Abstract

Introduction: Hirsutism is a condition defined as excessive male-pattern hair growth in females most commonly caused by hyperandrogenism. Polycystic ovary syndrome (PCOS), non-classic adrenal hyperplasia (NCAH) due to 21-hydroxylase deficiency and androgen-secreting tumors represent causes of androgen excess. Common features such as hirsutism, polycystic ovaries, oligomenorrhea or amenorrhea, and insulin resistance make it hard to distinguish between the first two conditions.

Material and methods: A retrospective study was conducted at "Louis Turcanu" Children's Clinical and Emergency Hospital over two years. Twenty-three female patients admitted to the Endocrinology Department for hirsutism were included. Patient history and physical examination, blood sampling, ovarian sonography, oral glucose tolerance tests were performed in all patients. Insulin resistance (IR) was assessed by the homeostasis model assessment (HOMA).

Results and discussions: Sixteen patients were diagnosed with PCOS according to the Rotterdam criteria, and seven patients were diagnosed with NCAH. Obesity, defined as a BMI at or above the 95th percentile, had a higher prevalence among PCOS patients compared to NCAH patients; 52.9% obese PCOS patients compared to 33.3% obese NCAH patients. IR was diagnosed in 56.2% of PCOS patients. Oligomenorrhea was more frequent among pubertal patients with PCOS (31.2 %) compared to NCAH (14.2%). The highest mean DHEA levels were found in NCAH patients, 15.55 ± 7.81, 95% confidence interval [CI] compared to 12.74 ± 6.027, 95% CI in PCOS patients. Basal 17-hydroxyprogesterone levels >2 ng/mL were more prevalent in the NCAH group, whereas an LH/FSH ratio > 2 was predominant among PCOS patients.

Conclusions: Differentiating between PCOS and NCAH remains a diagnostic challenge. Basal 17-hydroxyprogesterone levels >2 ng/mL and LH/FSH ratio > 2 are useful markers in diagnosing the cause of androgen excess. Further, more extensive studies and genetic testing are needed.

Introduction

Hirsutism is defined as a condition of male-pattern hair growth in women. Most commonly, this excess hair growth is caused by increased levels of androgens (1,2). The most common causes of hyperandrogenism are polycystic ovary syndrome (PCOS), non-classic adrenal hyperplasia (NCAH) due to 21-hydroxylase deficiency and androgen-secreting tumors (3-5). Differentiating between PCOS and NCAH is difficult because of common features such as hirsutism, polycystic ovaries, oligomenorrhea or amenorrhea, and insulin resistance (6,7).

Aim of the study:

To determine which clinical features and hormonal parameters can be used in order to distinguish PCOS from NCAH, in hirsute female patients.

Material and methods:

- ✓retrospective observational study
- ✓period : ***2 years*** (January 1st 2017 - December 31th 2018)
- ✓*Endocrinology Department* - "Louis Turcanu" Children's Clinical and Emergency Hospital *Timisoara*
- ✓initial batch: ***23 female patients***
- ✓study groups:
  - ✓ 16 patients diagnosed with PCOS
  - ✓ 7 patients diagnosed with NCAH

Results and Discussions

Table1: Descriptive statistics of the studied groups

Group n	**PCOS** 16	**NCAH** 7
Clinical features		
Age (years)	13.18 ± 2.88	15.57 ± 1.81
BMI (kg/m²)	24.67 ± 4.92	22.74 ± 4.78
Acanthosis nigricans	5/16	0/7
Ferriman-Gallwey score	15.87 ± 3.20	16.85 ± 4.87
Oligomenorrhea	5/16	1/7
Polycystic ovaries	16/16	2/7
Hormonal parameters		
DHEA ng/mL	15.55 ± 7.81	15.55 ± 7.81
17-OHP > 2 ng/mL	4/16	4/7
LH/FSH >2	9/16	0/7

The descriptive statistics of the studied groups are shown in Table 1. PCOS patients had higher BMI scores compared to NCAH patients, with a mean of 24.67 ± 4.92 kg/m² **and** 22.74 ± 4.78 kg/m², respectively. The mean Ferriman-Gallwey score was slightly higher in patients with NCAH (16.85 ± 4.87), than in those with PCOS (15.87 ± 3.20). Acanthosis nigricans, oligomenorrhea and polycystic ovaries were more frequently observed among PCOS patients (5/16, 5/16 and 16/16) than in those with NCAH (0/7, 1/7 and 2/7). NCAH patients showed the highest mean DHEA levels, 15.55 ± 7.81 ng/mL, compared to 12.74 ± 6.027, in PCOS patients.

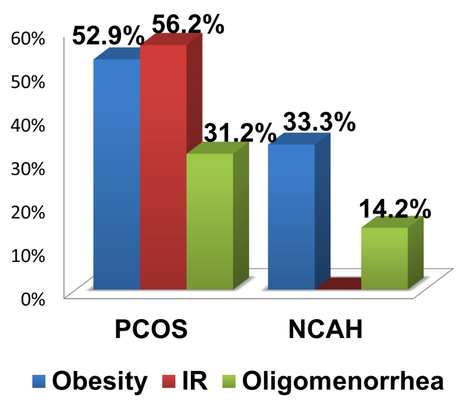

Fig.1: Percentage distribution of clinical features in the studied groups

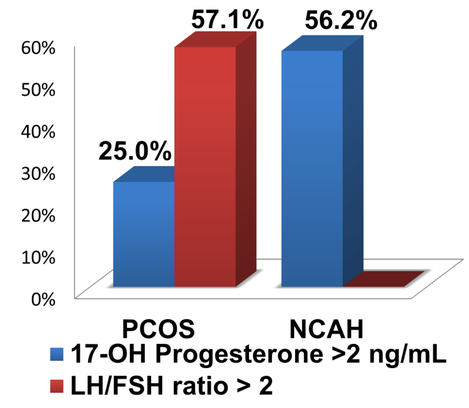

Fig.2: Percentage distribution of hormonal parameters in the studied groups

Obesity had a significantly higher prevalence among PCOS patients compared to NCAH patients (52.9% and 33.3 %). Oligomenorrhea was observed in 31.2 % of PCOS cases, double the precentage of NCAH patients with this clinical sign.

Basal 17-hydroxyprogesterone levels >2 ng/mL were more prevalent in the NCAH group, in 56.2% of cases, whereas an LH/FSH ratio > 2 was predominant among PCOS patients, 57.1% patients.

Conclusions

Differentiating between PCOS and NCAH remains a diagnostic challenge. Basal 17-hydroxyprogesterone levels >2 ng/mL and LH/FSH ratio > 2 are useful markers in diagnosing the cause of androgen excess. Further, more extensive studies and genetic testing are needed.

Bibliography

1.Hohl, Alexandre, Ronsoni, Marcelo Fernando, & Oliveira, Mônica de. (2014). Hirsutism: diagnosis and treatment. Arquivos Brasileiros de Endocrinologia & Metabologia, 58(2), 97-107. https://dx.doi.org/10.1590/0004-2730000002923

2.Brodell, L. A. and M. G. Mercurio (2010). "Hirsutism: Diagnosis and management." Gend Med 7(2): 79-87, doi: 10.1016/j.genm.2010.04.002.

3.Azziz R, Hincapie LA, Knochenhauer ES, Dewailly D, Fox L, Boots LR. Screening for 21-hydroxylase-deficient nonclassic adrenal hyperplasia among hyperandrogenic women: a prospective study. Fertil Steril 1999;72:915 –925.

4.Azziz R, Carmina E, Dewailly D, Diamanti-Kandarakis E, Escobar-Morreale HF, Futterweit W, Janssen OE, Legro RS, Norman RJ, Taylor AE et al. Position statement: criteria for defining polycystic ovary syndrome as a predominantly hyperandrogenic syndrome: an Androgen Excess Society guideline. J Clin Endocrinol Metab 2006;91:4237 –4245

5.Escobar-Morreale, H. F., et al. (2012). "Epidemiology, diagnosis and management of hirsutism: a consensus statement by the Androgen Excess and Polycystic Ovary Syndrome Society." Hum Reprod Update 18(2): 146-170, DOI: 10.1093/humupd/dmr042.

6.Papadakis, G., et al. (2019). "Polycystic Ovary Syndrome and NC-CAH: Distinct Characteristics and Common Findings. A Systematic Review." Frontiers in Endocrinology 10(388) https://doi.org/10.3389/fendo.2019.00388.

7.Pignatelli, D. (2013). "Non-classic adrenal hyperplasia due to the deficiency of 21-hydroxylase and its relation to polycystic ovarian syndrome." Front Horm Res 40: 158-170.

58ESPE P3-303

Sex differentiation, gonads and gynaecology or sex endocrinology

Teofana Otilia Bizerea-Moga

Poster presented at: ESPE 2019 VIENNA

PosterSessionOnline.com